GP6 gene

glycoprotein VI platelet

Normal Function

The *GP6* gene provides instructions for making a protein called glycoprotein VI (GPVI). GPVI is a receptor protein that is embedded in the outer membrane of blood cell fragments called platelets, which are an essential component of blood clots. Normally, blood clots protect the body after an injury by sealing off damaged blood vessels and preventing further blood loss.

Receptor proteins, like GPVI, have specific sites into which certain other proteins, called ligands, fit like keys into locks. Together, ligands and their receptors trigger signals that affect cell function. The main ligand for GPVI is a protein called collagen, which is found on blood vessel walls. In response to an injury that causes bleeding, the GPVI protein attaches (binds) to collagen, which begins clot formation and signals additional platelets to come together to increase the size of the clot. The GPVI protein can also bind to a protein called fibrin, which is the main protein that forms blood clots.

Health Conditions Related to Genetic Changes

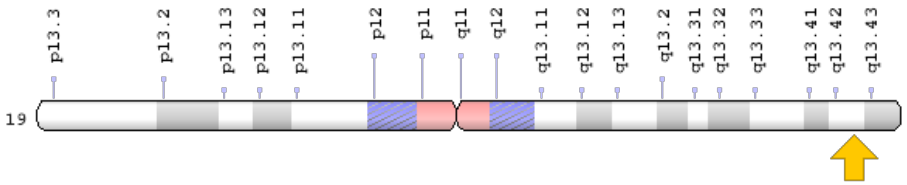
glycoprotein VI deficiency

At least five mutations in the *GP6* gene have been found to cause glycoprotein VI deficiency, a bleeding disorder associated with a decreased ability to form blood clots. The mutations lead to the production of no GPVI protein; an abnormally short, nonfunctional GPVI protein; or a protein that is less able to bind to collagen. Without GPVI binding to collagen, platelets cannot come together efficiently at the site of an injury to form a clot, leading to an increased risk of nosebleeds, abnormally heavy or prolonged bleeding following minor injury or surgery, or other bleeding problems associated with glycoprotein VI deficiency.

Chromosomal Location

Cytogenetic Location: 19q13.42, which is the long (q) arm of chromosome 19 at position 13.42

Molecular Location: base pairs 55,013,705 to 55,038,264 on chromosome 19 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- glycoprotein 6
- GPVI
- GPVI collagen receptor
- platelet collagen receptor
- platelet glycoprotein VI
- platelet membrane glycoprotein VI

Additional Information & Resources

Educational Resources

- Platelet-Vessel Wall Interactions in Hemostasis and Thrombosis (2010): Platelet Adhesion Molecules
  https://www.ncbi.nlm.nih.gov/books/NBK53455/#s2.3

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28GP6%5BTIAB%5D%29+OR+%28glycoprotein+VI+platelet%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- GLYCOPROTEIN VI, PLATELET
  http://omim.org/entry/605546

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=GP6%5Bgene%5D
- HGNC Gene Family: Immunoglobulin like domain containing
  http://www.genenames.org/cgi-bin/genefamilies/set/594
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=14388
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/51206
- UniProt
  http://www.uniprot.org/uniprot/Q9HCN6

Sources for This Summary

- Arthur JF, Dunkley S, Andrews RK. Platelet glycoprotein VI-related clinical defects. Br J Haematol. 2007 Nov;139(3):363-72. Review.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/17910626
- Dumont B, Lasne D, Rothschild C, Bouabdelli M, Ollivier V, Oudin C, Ajzenberg N, Grandchamp B, Jandrot-Perrus M. Absence of collagen-induced platelet activation caused by compound heterozygous GPVI mutations. Blood. 2009 Aug 27;114(9):1900-3. doi: 10.1182/blood-2009-03-213504.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19549989
- OMIM: GLYCOPROTEIN VI, PLATELET
  http://omim.org/entry/605546
- Hermans C, Wittevrongel C, Thys C, Smethurst PA, Van Geet C, Freson K. A compound heterozygous mutation in glycoprotein VI in a patient with a bleeding disorder. J Thromb Haemost. 2009 Aug;7(8):1356-63. doi: 10.1111/j.1538-7836.2009.03520.x.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/19552682
- Matus V, Valenzuela G, Sáez CG, Hidalgo P, Lagos M, Aranda E, Panes O, Pereira J, Pillois X, Nurden AT, Mezzano D. An adenine insertion in exon 6 of human GP6 generates a truncated protein associated with a bleeding disorder in four Chilean families. J Thromb Haemost. 2013 Sep;11(9):1751-9. doi: 10.1111/jth.12334.
  Citation on PubMed: https://www.ncbi.nlm.nih.gov/pubmed/23815599

Reviewed: April 2017
Published: April 25, 2017

Lister Hill National Center for Biomedical Communications
U.S. National Library of Medicine
National Institutes of Health
Department of Health & Human Services